



Stargardt macular degeneration

Stargardt macular degeneration is a genetic eye disorder that causes progressive vision loss. This disorder affects the retina, the specialized light-sensitive tissue that lines the back of the eye. Specifically, Stargardt macular degeneration affects a small area near the center of the retina called the macula. The macula is responsible for sharp central vision, which is needed for detailed tasks such as reading, driving, and recognizing faces. In most people with Stargardt macular degeneration, a fatty yellow pigment (lipofuscin) builds up in cells underlying the macula. Over time, the abnormal accumulation of this substance can damage cells that are critical for clear central vision. In addition to central vision loss, people with Stargardt macular degeneration have problems with night vision that can make it difficult to navigate in low light. Some affected individuals also have impaired color vision. The signs and symptoms of Stargardt macular degeneration typically appear in late childhood to early adulthood and worsen over time.

Frequency

Stargardt macular degeneration is the most common form of juvenile macular degeneration, the signs and symptoms of which begin in childhood. The estimated prevalence of Stargardt macular degeneration is 1 in 8,000 to 10,000 individuals.

Genetic Changes

In most cases, Stargardt macular degeneration is caused by mutations in the *ABCA4* gene. Less often, mutations in the *ELOVL4* gene cause this condition. The *ABCA4* and *ELOVL4* genes provide instructions for making proteins that are found in light-sensing (photoreceptor) cells in the retina.

The *ABCA4* protein transports potentially toxic substances out of photoreceptor cells. These substances form after phototransduction, the process by which light entering the eye is converted into electrical signals that are transmitted to the brain. Mutations in the *ABCA4* gene prevent the *ABCA4* protein from removing toxic byproducts from photoreceptor cells. These toxic substances build up and form lipofuscin in the photoreceptor cells and the surrounding cells of the retina, eventually causing cell death. Loss of cells in the retina causes the progressive vision loss characteristic of Stargardt macular degeneration.

The *ELOVL4* protein plays a role in making a group of fats called very long-chain fatty acids. The *ELOVL4* protein is primarily active (expressed) in the retina, but is also expressed in the brain and skin. The function of very long-chain fatty acids within the retina is unknown. Mutations in the *ELOVL4* gene lead to the formation of *ELOVL4*

protein clumps (aggregates) that build up and may interfere with retinal cell functions, ultimately leading to cell death.

Inheritance Pattern

Stargardt macular degeneration can have different inheritance patterns.

When mutations in the *ABCA4* gene cause this condition, it is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

When this condition is caused by mutations in the *ELOVL4* gene, it is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

Other Names for This Condition

- juvenile macular degeneration
- macular dystrophy with flecks, type 1
- Stargardt disease
- STGD

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Stargardt disease 1
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1855465/>
- Genetic Testing Registry: Stargardt Disease 3
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1838644/>
- Genetic Testing Registry: Stargardt disease 4
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1863534/>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>

- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

MedlinePlus

- Health Topic: Macular Degeneration
<https://medlineplus.gov/maculardegeneration.html>
- Health Topic: Retinal Disorders
<https://medlineplus.gov/retinaldisorders.html>

Genetic and Rare Diseases Information Center

- Stargardt disease
<https://rarediseases.info.nih.gov/diseases/181/stargardt-disease>

Additional NIH Resources

- National Eye Institute: Diagram of the Eye
<https://nei.nih.gov/health/eyediagram/>

Educational Resources

- Disease InfoSearch: Stargardt disease
<http://www.diseaseinfosearch.org/Stargardt+disease/6861>
- MalaCards: stargardt macular degeneration
http://www.malacards.org/card/stargardt_macular_degeneration
- My46 Trait Profile
<https://www.my46.org/trait-document?trait=Stargardt%20disease&type=profile>
- Orphanet: Stargardt disease
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=827

Patient Support and Advocacy Resources

- American Foundation for the Blind
<http://www.afb.org/>
- National Organization for Rare Disorders (NORD): Macular Degeneration
<https://rarediseases.org/rare-diseases/macular-degeneration/>
- Prevent Blindness America
<http://www.preventblindness.org/>

- The Foundation Fighting Blindness
<http://fffb.ca/wp-content/uploads/2015/12/stargardt.pdf>
- University of Kansas Medical Center Resource List: Retinal Degeneration
<http://www.kumc.edu/gec/support/retinal.html>

ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?term=%22Stargardt+macular+degeneration%22+%5BDISEASE%5D+OR+NCT00254605+%5BID-NUMBER%5D+OR+NCT01278277+%5BID-NUMBER%5D+OR+NCT00060749+%5BID-NUMBER%5D+OR+NCT00346853+%5BID-NUMBER%5D+OR+NCT00420602+%5BID-NUMBER%5D>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28Macular+Degeneration%5BMAJR%5D%29+AND+%28%28stargardt+macular+degeneration%5BTIAB%5D%29+OR+%28stargardt+disease%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- STARGARDT DISEASE 1
<http://omim.org/entry/248200>
- STARGARDT DISEASE 3
<http://omim.org/entry/600110>

Sources for This Summary

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